

Editorial

During the last decade, research addressing either the underlying cause of rare genetic disorders or the genetic components of complex diseases has tremendously elucidated our understanding of disease causing mechanisms in all medical specialties. The enormous technical progress enabling these developments has also revolutionized the diagnostic yield in previously clinically obscure conditions. Karyotyping by means of microscopic analysis of stained chromosomes for examples is about to be replaced by molecular karyotyping using microarrays with millions of probes to detect minimal aberrations far beyond the capability of the human eye. Sequencing of genes is no longer limited to the detection of the disease causing mutation in clinically unambiguous cases for reproductive management, but has become an indispensable tool for establishing a diagnosis especially in ambiguous cases. In the light of this technical revolution, genetic syndromology, the art of recognizing a certain condition through a more or less characteristic and often very variable combination of clinical signs and the knowledge about the natural history of these syndromes was written off by many geneticists. Along with this attitude reports of molecular genetic findings especially in high ranking journals often neglected clinical detail. However, the previously unimagined flood of information arising through very high resolution karyotyping and anticipated at the dawn of massive parallel sequencing, reinstructs us about the importance of the thorough clinical observation as a prerequisite to turn molecular data into useful medical information.

This new journal was therefore introduced to emphasize the relation between genotype and phenotype with special focus on translating molecular findings into medical practice.

Molecular Syndromology publishes research articles, short reports and reviews on common and rare genetic syndromes, aiming to increase clinical understanding through molecular insights. Topics of particular interest are the molecular basis of genetic syndromes, genotype-phenotype correlation, natural history, strategies in disease management and novel therapeutic approaches based on molecular findings. Research on model systems is also welcome, when it is obviously relevant to medical genetics. With high-quality reviews and special issues devoted to current topics the journal aims to facilitate early transfer of research findings to a clinical setting while also stimulating further research on medically relevant questions. The feature of 'Calls for Collaborative and Interactive Research Projects' enables joining of forces in very rare disorders. The journal targets not only medical geneticists and basic biomedical researchers, but also clinicians dealing with genetic syndromes. With four Associate Editors from three continents and a broad international Editorial Board the journal welcomes submissions covering the latest research from around the world.

I therefore trust that you will find *Molecular Syndromology* a useful and trustworthy companion in your medical research and clinical practice.

Anita Rauch